

Genetic services and testing in the Philippines

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Introduction

The Philippines is a developing middle-income country in Southeast Asia and is regarded as one of the fastest growing economies in the region. It became a Spanish colony for more than three centuries (16th to 19th century), after which the Americans took over until the Japanese came in the 1940s during World War II. With the help of the Americans, the Filipinos regained their freedom. The country became a republic and achieved sovereignty in 1946. With a very long history of colonizations and intermarriages, the Filipinos are said to be a mixture of different races, including Malay, Chinese, Spanish, Negrito and American. Most of the people in the country are Tagalog, and the rest belong to other ethnic groups such as Cebuano, Ilocano, Bisaya, Hiligaynon Ilonggo, Bicolano and Waray. Majority of Filipinos are Catholics, but Muslims and other Christian and non-Christian sects also play important roles in the society. The official languages used in the country are Filipino (Tagalog) and English (World Factbook 2011; Dolan 1991).

In general, health indices in the country show an improving trend due to the programs of the Department of Health (DOH) and local government units (LGUs). However, despite this progress, issues of poverty and compromised access to health services in remote areas

continue as pressing concerns. Additionally, the country's health expenditure per capita remains on the low side compared to other countries in the region (UNDP 2009; UNESCO 2009).

Delivery of medical genetic services remains to be a challenge in both private and public sectors. Since infectious diseases are still in the top ten causes of infant mortality and morbidity, limited attention is given to congenital anomalies and other genetic diseases, despite the fact that congenital anomalies are included in the top ten causes of infant mortality (DOH 2006). This is aggravated by the lack of geneticists and genetic counsellors to serve the patients that are spread out in numerous islands.

Demography and health indicators

Located in Southeast Asia, the Philippines is an archipelago covering an area of 340,574.7 km² (Fig. 1). The three major island groups—Luzon, Visayas and Mindanao—are made up of a total of 7,107 islands (NSO 2009).

The country is the 12th most populous country in the world, seventh in Asia and second in Southeast Asia. As of 2010, the population was reported at 94 million, with a population density of 276 persons/km². The annual growth rate is around 2 %, with 0.4 % of the population comprised of immigrants. Majority of the population are in the rural areas (IMF 2011; UNDP 2009; NSO 2009).

Life expectancy is an average of 71 years for both sexes (74 years for women, 67 years for men) and has improved since the 1990s. The healthy life expectancy also has a similar trend at an average of 64 years for both sexes (64 years for women, 59 years for men). Furthermore, adult mortality rates have decreased over the years for both sexes

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Fig. 1 Map of the Philippine archipelago at http://www.freeusandworldmaps.com/images/Countries_Maps/AsiaCountries/Philippines/Philipp (accessed June 5, 2012)

(men higher than women). Maternal mortality ratio remains high at 230 per 100,000 livebirths in 2005 (WHO 2009).

There is a decreasing trend of fertility rate at around three children per woman, which is projected to continue until the following years (NSO 2008; 2009). Approximately 15 % of deliveries are born to women aged 35 years and above (NSO 2009).

The neonatal mortality rates (15 per 1,000 livebirths in 2008), infant mortality rates (25 per 1,000 livebirths in 2008), and under-5 mortality rates (34 per 1,000 livebirths in 2008) have declined since the 1990s due to the improved maternal health care (NSO 1998, 2003, 2009). Additionally, there is an increase in births delivered in health facilities but the percentage of home births remains high at 60 % (NSO 1998; 2003; 2009). Among pregnant women, 88 % received at least one antenatal care visit from skilled health workers with a concomitant decrease in antenatal care from a traditional birth attendant (*hilot*). This was associated with an improved percentage of deliveries attended by skilled health personnel (WHO 2009; NSO 1998; 2003; 2008).

Health expenditure and financing

The gross national income of the Philippines as of 2008 is US \$35.6 billion (NSO 2009). In 2005, total health expenditure went up by 9.4 %, at US\$4.2 billion, with the following breakdown of sources: government (28.7 %) shared by national (15.8 %) and local government (12.9 %); social insurance (11 %); private sources (59.1 %) shared by the out-of-pocket expenses (48.4 %), and private insurance (2.4 %); private health plans (3.9 %); employer-based plans (3.2 %); private schools (1.2 %); and others (1.2 %). The almost 50 % out-of-pocket expenditure is a burden to the families and this is way above the target of 20 % (DOH 1999; PNHA 2005).

The health system is guided by the “Health Sector Reform Agenda (HSRA) Philippines 1999–2004,” a comprehensive plan for changing the Philippine health care system. It describes the policies, public investments, and organizational changes needed to improve the way health care is delivered, regulated, and financed in the country. Specifically, it seeks to undertake the following: (1) provide fiscal autonomy to government hospitals; (2) secure funding for priority public health programs; (3) promote the development of local health systems and ensure their effective performance; (4) expand the coverage of the National Health Insurance Program (NHIP); and (5) strengthen the capacities of health regulatory agencies, i.e., Bureau of Food and Drug (BFAD), Bureau of Health Devices and Technology (BHDT), Bureau of Health Facilities and Services (BHFS), and Bureau of Quarantine and International Health Surveillance (BQIHS). Ultimately, HSRA aims to improve the health status of the people through greater and more

effective coverage of national and local public health programs; increase access to health services especially by the poor; and reduce financial burden on individual families. It was envisioned to be the catalyst that will bring the country towards the realization of a shared vision of health for all Filipinos (DOH 1999).

The share of health expenditure to gross domestic product (GDP) is still below the 5 % standard set by the WHO for developing countries. However, the share of health expenditure to gross national product (GNP) remained at 3.1 %, which is within the National Objectives for Health target of 3–4 % (PNHA 2005).

Congenital and genetic disease burden

There are no national surveys or registries on congenital defects and genetic disorders in the Philippines. The estimated birth defects prevalence is 52.9 per 1,000 livebirths (Christianson et al. 2006).

The annual registry of causes of infant mortality lists congenital malformations of the heart and other congenital malformations in the top ten since the 1960s. In 2006, 1.4 infant deaths per 1,000 livebirths and 8.72 under-5 deaths per 100,000 population, were due to congenital disorders (Padilla et al. 2011a; DOH 2006).

In 1999, the Institute of Human Genetics, National Institutes of Health (IHG-NIH), University of the Philippines (UP) Manila in partnership with DOH conducted a pilot project of the Birth Defects Registry. This project involved examination of 191,576 newborns in 79 hospitals which revealed a total of 1,240 birth defect cases. This project had limitations: (1) it was not integrated into the programs of DOH, and (2) it was limited to hospital-born babies (Padilla et al. 2003). Since there was no systematic surveillance for congenital anomalies, there were no organized programs to cater to the needs of these patients. With neural tube defects in the top ten birth defects (sixth in rank), awareness campaigns on the importance of folic acid and discussions on folic acid supplementation were carried out by the DOH. Folic acid was added in the vitamins for mothers. However, there was no aggressive campaign on the importance of folic acid in-between pregnancies and especially prior to conception.

The birth defects registry of 79 hospitals in 1999 (Padilla et al. 2003) and a birth defects registry at the Philippine General Hospital (PGH) (Padilla et al. 2011b), the biggest government tertiary hospital in the Philippines, have both documented that cleft lip and cleft palate are among the most common birth defects in the country. To respond to the high number of patients with cleft lip and palate, several missions (both local and international) are conducted year round to perform surgery on these patients.

Table 1 Prevalence of congenital disorders by cause

Cause	Estimated prevalence (per 1,000 livebirths)
Dominant single-gene disorders	7
Recessive single gene disorders	2.3
X-linked recessive single gene disorders	1.3
Chromosomal disorders	4.2
Malformations	63.9

Source: Christianson et al. (2006) March of Dimes (MoD) Global Report on Births Defects, 2006: The Hidden Toll of Dying and Disabled Children

The March of Dimes (MoD), USA, has involved the Philippines in the Global Network for Maternal and Infant Health (GNMIH), whose goal is to reduce mortality and disability from birth defects and preterm births in lower- and middle-income countries. Together with China and Lebanon, the Philippines embarked on birth defects surveillance projects to identify and help affected patients. Activities under GNMIH include: a prospective birth defects surveillance, prevention of birth defects campaigns, engaging the youth in GNMIH activities [Volunteer Youth Leaders for Health (VYLH)] and preconception activities for the prevention of birth defects (Padilla et al. 2011a; MoD 2012). Likewise, the birth defects registry under GNMIH showed cleft lip and cleft palate to be in the top ten of birth defects. Oral clefts are common birth defects of complex genetic and environmental etiologies. A review of cases ascertained through surgical cleft missions revealed an incidence of one in 2,367 livebirths (Padilla et al. 2008). Another study by Murray et al. reported an incidence of 1:500, using records from an 8-year period at a government hospital in Negros, the Corazon Locsin Montelibano Memorial Regional Hospital (Murray et al. 1997).

The MoD reported the estimated prevalence of congenital disorders in 2006 as listed in Table 1 (Christianson et al. 2006). Table 2 shows the data of the National Newborn Screening Program (NSRC 2011). Table 3 presents data on Filipino newborns who underwent expanded newborn screening in California, USA (personal communication with Fred Lowry and Lisa Feuchtbaum of the California Newborn Screening Program 2011). The observed differences in the prevalence of some disorders may be due to the different sample sizes. Table 4 presents data from the Biochemical Genetics Laboratory of IHG-NIH from 1999 to 2011.

Availability of genetic services

Several genetic laboratory services were introduced in the 1980s. Genetic counselling services were offered by four (4) units: (1) Department of Pediatrics, PGH; (2) Department of

Obstetrics and Gynecology, PGH; (3) Department of Internal Medicine, PGH; and (4) Department of Pathology and Microbiology in the University of the East–Ramon Magsaysay Memorial Medical Center. The field of genetics slowed down with the untimely demise of two geneticists and the retirement of another (Briones et al. 1984; Padilla 2008).

Genetic services at the University of the Philippines

Re-emergence of clinical genetics was seen in the 1990s with the establishment of the Medical Genetics Unit (MGU). It was closely linked with the Royal Alexandra Hospital for Children, Sydney, Australia (later renamed The Children's Hospital Westmead), where clinical and laboratory personnel pursued training.

The MGU started as a special project under the Office of the Dean of the UP College of Medicine, in recognition of the need to provide comprehensive genetic services in the country. Initially housed at the Department of Pediatrics, the Dysmorphology Clinic (later renamed the Clinical Genetics Clinic) was opened at the PGH Out-Patient Department in 1991. Cytogenetic laboratory services were opened the same year at the Pediatric Research Laboratory. The Newborn Screening (NBS) Laboratory was opened in 1997. In 1999, IHG was established as one of the component institutes of the NIH. The Molecular Genetics Laboratory and the Biochemical Genetics Laboratory were opened in 1999 and 2001, respectively, as part of the expansion of IHG. Subsequently, all laboratory services were housed at the NIH (Padilla 2008), while the clinical services (in-patient and out-patient) remained at the PGH Department of Pediatrics.

Today, the IHG-NIH is the largest provider of genetic services in the Philippines. It has the following units: clinical genetics unit, cytogenetics unit, newborn screening center, molecular genetics unit and biochemical genetics unit. Aside from genetic services, the units are also involved in research and advocacy (Padilla 2008; Padilla et al. 2012).

Table 2 Prevalence of disorders among Filipino newborns in the Newborn Screening Program from 1996 to 2011^a

Conditions	Number of cases	Prevalence
Congenital adrenal hyperplasia	293	1: 10,604
Congenital hypothyroidism	1,034	1: 3,004
Galactosemia (classical and non-classical)	34	1: 91,380
Galactosemia, variant form	101	1: 30,762
Phenylketonuria (classical and mild)	17	1: 182,761
Hyperphenylalanemia	16	1: 194,183
Glucose-6-phosphate dehydrogenase deficiency	60,204	1: 50

^a Total of 3,106,938 newborns screened (www.newbornscreening.ph)

Table 3 Prevalence of disorders among Filipino newborns in a California, USA, Newborn Screening Laboratory, from 2005 to 2011^a

Conditions	Number of cases	Prevalence
Endocrinologic Disorders		
Congenital adrenal hyperplasia (classical salt wasting)	1	1:111,127
Primary congenital hypothyroidism	45	1: 2,469
Transient hypothyroidism	4	1: 27,782
Variant hypothyroidism	1	1:111,127
Hemoglobinopathies		
α -Thalassemia major	5	1: 22,225
β -Thalassemia major	1	1: 111,127
Hb E/Beta + thalassemia	1	1: 111,127
Hb H disease	93	1: 1,195
Hb H/constant spring disease	2	1: 55,564
Hb variant/beta + thalassemia	1	1: 111,127
Homozygous EE	3	1: 37,042
Sickle C disease (Hb S/C disease)	2	1: 55,564
Sickle cell anemia (Hb S/S disease)	1	1:111,127
Amino acid disorders		
Phenylketonuria (PKU)	4	1: 27,782
Variant hyperphenylalaninemia	1	1:111,127
Maple syrup urine disease (MSUD)	1	1:111,127
Organic acid disorders		
Methylmalonic acidemia (MMA) (mut 0)	3	1: 37,042
Methylmalonic acidemia (MMA) (mut -)	2	1: 55,564
β -Ketothiolase deficiency (BKT)	1	1: 111,127
Isobutyryl-CoA dehydrogenase deficiency (IBDHD)	1	1:111,127
Fatty acid oxidation disorders		
Medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)	2	1:55,564
Short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)	3	1: 37,042
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)	3	1: 37,042
Other fatty acid oxidation disorder	2	1: 55,564
Others		
Partial BD deficiency	1	1:111,127
CFTR-related metabolic syndrome (CRMS)	5	1:22,225
Cystic fibrosis	5	1: 22,225
Classical galactosemia	1	1: 111,127
Duarte galactosemia (D/G)	2	1: 55,564
Other disorders	2	1: 55,564

^a Total of 111,127 newborns screened

Clinical Genetics Unit

The Clinical Genetics Unit of the IHG-NIH provides comprehensive clinical services to families and individuals with, or at risk for, an inherited disease. It offers information and genetic counselling to families to allow individuals the

Table 4 Number of patients with metabolic disorders at the Biochemical Genetics Laboratory, IHG-NIH from 1999 to 2011

Condition	Number of cases
Amino acid disorders	
Maple syrup urine disease	101
Phenylketonuria	17
Hyperphenylalaninemia	10
Heterozygous cystinuria	9
6-Pyruvoyltetrahydropterin synthase deficiency	4
Homozygous cystinuria	3
Homocystinuria	1
Tyrosinemia	1
Urea cycle disorders	
Carbamoyl phosphate synthase deficiency	4
Ornithine transcarbamoylase deficiency	3
Citrullinemia	1
Arginosuccinate lyase deficiency syndrome	1
Organic acid disorders	
Methylmalonic aciduria	15
Glutaric aciduria type I	2
Holocarboxylase synthase deficiency	1
Mitochondrial diseases	
Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke (MELAS) syndrome	2
Respiratory chain complex deficiency	1
Peroxisomal Disorders	
Adrenoleukodystrophy	11
Lysosomal Storage Disorders	
Mucopolysaccharidosis	36
Gaucher disease	10
Pompe disease	3
Multiple sulfatase deficiency	1
Niemann–Pick disease	1
Mucopolipidosis	1
Tay–Sach's disease	1
Purine-pyrimidine Metabolism Disorders	
Lesch–Nyhan disease	1
Others	
Non-classical galactosemia	18
Classical galactosemia	5
Lowe syndrome	1
Menkes disease	1

widest possible choice of options, within the resources available, when making decisions related to their genetic risk. Clinical genetics services are rendered at the PGH Department of Pediatrics, a 1,500-bed tertiary government hospital serving as the national university teaching hospital of the UP Manila Health Sciences campus. Services are provided by geneticists,

genetics fellows-in-training, pediatric residents rotating at the Clinical Genetics Unit, and nurses.

The activities are: (1) general genetics out-patient service (six half-day clinics per month) which receives intra- and inter-hospital and private physician referrals; (2) genetic metabolic clinic (two half-day clinics per month) to take care of patients with inborn errors of metabolism, including enzyme replacement therapy for Gaucher disease and Pompe disease; (3) mucopolysaccharidoses (MPS) clinic (every 6 months) where patients are seen by a multi-disciplinary team (Padilla 2008; PSOD 2006; Padilla et al. 2012); and (4) support group meetings.

The top diagnoses made in the out-patient clinics and in-patient referrals are listed in Table 5 (IHG-NIH census, 2007–2008, unpublished).

Clinical genetic services became available in Cebu City and Davao City in 2004 and 2008, respectively. Clinical genetic services are also available in the private clinics of the clinical geneticists. Genetic work-up is coordinated with IHG-NIH and other overseas laboratories.

Cytogenetics Unit

Initially established under MGU in 1991, the Cytogenetics Unit became part of IHG-NIH. The unit performs routine karyotyping of peripheral blood cells, cord blood, solid tissues and bone marrow for patients with multiple birth defects, mental retardation, abnormal sexual development, for couples with infertility or multiple miscarriages, and for patients with malignancies and hematologic disorders. It also offers high-resolution banding and *fluorescence-in-situ hybridization* (FISH) analysis for testing other chromosomal disorders and

Table 5 Top diagnoses for out-patient genetics clinic consultations and in-patient referrals, Department of Pediatrics, PGH

Source of consults/ referrals	Top diagnoses
Out-patient clinic consultations	Down syndrome
	Maple syrup urine disease (MSUD)
	Non-syndromic global developmental delay/ mental retardation
	Phenylketonuria (PKU)
In-patient referrals	Multiple congenital anomalies, unspecified
	Down syndrome
	MSUD
	Multiple congenital anomalies, unspecified
	VACTERL association
	Failure to thrive

cancers. Chromosomal microarray analysis for selected clinical cases will be offered soon.

Newborn Screening Center

The Newborn Screening Center (NSC-NIH) administers both laboratory and follow-up services. In addition to laboratory testing, the NSC advises health care providers about appropriate diagnostic and treatment follow-up for abnormal screening results, provides long term follow-up for confirmed patients to evaluate screening outcomes, and provides training and education to health professionals and the general public. A total of four NSCs, which are strategically located, provide laboratory and follow-up services throughout the country: NSC-NIH, NSC-Central Luzon, NSC-Visayas, and NSC-Mindanao (NSRC 2011).

Molecular Genetics Unit

The Molecular Genetics Unit of IHG-NIH conducts interdisciplinary researches in collaboration with both local and international academic clinicians and scientists with specialist groups studying the genetics of monogenic and complex genetic conditions in the Filipino population. Using molecular genetics techniques, new diagnostic tools are applied for the conditions seen in the clinics to understand how these genes cause disease (Padilla 2008; Padilla et al. 2012). Molecular-based projects are closely linked to the Philippine Genome Center (PGC 2011). It has a DNA sequencing facility, a microarray facility and a bio-bank facility. For tests that are not available in the country, IHG-NIH facilitates the DNA extraction and the transport of samples to overseas laboratories, primarily in the US and Australia. Pre-genetic testing and post-genetic testing counselling are provided by the geneticists.

Biochemical Genetics Unit

The Biochemical Genetics Unit IHG-NIH, offers expert diagnostic testing and provides physician-assisted consultative services to clinicians in order to provide accurate diagnosis and appropriate management of inherited metabolic disorders. The array of biochemical tests includes urine metabolic screen by high voltage electrophoresis, quantitative amino acid analysis by high performance liquid chromatography and organic acid analysis by gas chromatography-mass spectrometry. It works closely with the Biochemical Genetics Section of The Children's Hospital Westmead, Sydney, Australia (Padilla 2008). It will serve as a reference laboratory for the expanded newborn screening program that will cover additional amino acid and organic acid disorders.

Study groups

The IHG faculty participates in the following study groups: Diabetes Study Group with endocrinologists; Cardiogenomics Study Group with cardiologists; Philippine Cancer Genetics Study Group with medical and surgical oncologists, gastroenterologists, pulmonologists, and otorhinolaryngologists; Philippine Autism Study Group with developmental pediatricians; Philippine Thalassemia Research Study Group with adult and pediatric hematologists; Philippine Kawasaki Disease Study Group with cardiologists, infectious disease specialists, and rheumatologists; Philippine Subacute Sclerosing Panencephalitis (SSPE) Study Group with neurologists; Philippine Birth Defects Surveillance Study Group with pediatricians, obstetricians, midwives and nurses; Philippine Oral Cleft Study Group with reconstructive surgeons and otorhinolaryngologists; and Subphenotyping and Genetics in Oral-facial Cleft in Families Study Group with UP Manila speech therapists and geneticists from University of Iowa and University of Pittsburgh.

Prenatal diagnosis and termination of pregnancy

Prenatal diagnosis is practiced in a very limited way. The most extensively used prenatal diagnostic procedure is prenatal ultrasonography, which utilizes two-dimensional ultrasound for congenital anomaly screening during the second trimester of pregnancy. This is complemented by fetal echocardiography in cases of congenital heart defects detected after a congenital anomaly scan. First trimester ultrasound screening has been introduced in some urban medical centers. More advanced techniques of three- and four-dimensional ultrasonography and color-Doppler ultrasound have recently gained popularity. Maternal serum screen, whether using single or multiple markers, as a non-invasive form of prenatal diagnosis, has not been customarily offered in the country (Cutiongco-de la Paz 2006).

Amniocentesis is offered only for anticipatory guidance for obstetricians and pediatricians for a problematic pregnancy or preparation for the birth of a child with birth defects. Chorionic villi sampling has not been normally offered in any institution. Termination of pregnancy is not allowed by law in the country (Cutiongco-de la Paz 2006).

Preconception care

There is no formal preconception care program in the Philippines. There are some preventive programs in place, such as food fortification and supplementation: (1) salt iodization to reduce the risk of iodine deficiency disorders — Republic Act No. 8172 [An Act for Salt Iodization Nationwide (RA8172 1995)]; and (2) fortification with essential micronutrients (e.g., iron in rice; vitamin A in white flour, refined sugar,

and cooking oil) to reduce micronutrient deficiencies — Republic Act No. 8976 [The Philippine Food Fortification Act of 2000 (RA 8976 2000; AO 2003-1119; AO 2002-0096; AO 2010-0010)].

There is a voluntary immunization program for Rubella. *Iligtas sa Tigdas ang Pinas*, a Door-to-Door Measles–Rubella (MR) immunization campaign launched to cover all children 9 months up to below 8 years old. It was held last 1998, 2004, 2007 and 2011 towards Philippines' commitment to eliminate measles by 2012, the target year agreed upon with the other countries in the Western Pacific Region (Rubella Campaign 2011).

Presidential Proclamation No. 1188 declares every fourth week of January as Goiter Awareness Week (GAW). The Philippine Thyroid Council (PTC), a coalition of medical specialty societies, government and non-government organizations, professional associations and allied groups was created to spearhead the activity for the said event (Presidential Proclamation No. 1188 2006).

Disability services

Republic Act 9442, an act amending the Magna Carta for Persons with Disability (RA 7277) was passed on July 24, 2006. The passing of the new regulations secured the livelihood, health, economic and social well-being of those with disabilities. With the law, LGUs were called upon to establish a “Persons with Disabilities Affairs Office” (PDAO), which addresses specific needs in concentrated areas, such as accessibility, funding, ordinances and policies (Republic Act No. 9442 2006).

The National Council for Disability Affairs (NCDA) (formerly National Council for the Welfare of Disabled Persons [NCWDP]) is the national government agency mandated to formulate policies and coordinate the activities of all agencies, whether public or private, concerning disability issues and concerns. As such, the NCDA is the lead agency tasked to steer the course of program development for persons with disabilities and the delivery of services to the sector. The NCDA is tasked to monitor the implementation of several laws to ensure the protection of persons with disabilities' civil and political rights (NCDA 2011).

Other institutions with genetic services

There are other institutions (private and public) that are also providing genetic laboratory services. These include: (1) the Philippine Nuclear Research Institute (PNRI) — cytogenetics; (2) the Research and Biotechnology Division of St. Luke's Medical Center (SLMC) — cytogenetics and molecular genetics; (3) the Natural Science Research Institute (NSRI) UP Diliman — paternity testing and forensics; (4) the Philippine National Police (PNP) — forensics; 5) the

National Bureau of Investigation (NBI) — forensics; and 6) the National Kidney and Transplant Institute (NKTI) — cytogenetics. Clinical genetic services are provided in the private clinics of the clinical geneticists.

Population-based genetic screening programs

Newborn bloodspot screening

Newborn bloodspot screening (NBS) was introduced in the Philippines in 1996 by obstetricians and pediatricians from 24 private and government hospitals. NBS was integrated into the public health delivery system with the enactment of Republic Act 9288 or Newborn Screening Act of 2004. It institutionalized the “national comprehensive NBS System,” which shall ensure the following: (1) that every baby born in the Philippines is offered NBS; (2) the establishment and integration of a sustainable NBS System within the public health delivery system; (3) that all health practitioners are aware of the benefits of NBS and of their responsibilities in offering it; and (4) that all parents are aware of NBS and their responsibility in protecting their child from any of the disorders. The highlights of the law and its implementing rules and regulations are:

- DOH is the lead agency tasked with implementing this law.
- Any health practitioner, who delivers or assists in the delivery of a newborn in the Philippines shall, prior to delivery, inform parents or legal guardians of the newborns the availability, nature, and benefits of NBS.
- Health facilities shall integrate NBS in its delivery of health services.
- Creation of a Newborn Screening Reference Center (NSRC) at the NIH.
- Establishment and accreditation of NSCs equipped with an NBS laboratory and recall/follow-up program.
- Provision of NBS services as a requirement for licensing and accreditation at the DOH and the Philippine Health Insurance Corporation (PHIC).
- Inclusion of cost of NBS in insurance benefits.

Republic Act (RA) 9288 provided for the creation of the NSRC, an office that provides technical assistance to the DOH, and information and resources on NBS for the health professionals, public health community, consumers and government officials (Padilla et al. 2009a). Cost-benefit analysis of the national newborn screening program showed a benefit/cost ratio of 1.4 with net savings of US\$11.42M (Padilla et al. 2009b).

PHIC initially funded 90 % of the NBS fee (December 2006), but recently has issued a revised circular, funding 100 % of the newborn screening fee effective September 2011 (NSRC 2011).

From 1996 to December 2011, a total of 3,103,539 newborns have been screened, and the program has saved 60,204 patients. As of December 2011, the national NBS coverage is 42.0 %, a reflection of the success of the facility-based implementation since 40 % are born in the hospital. The remainder are born at home. Table 3 lists the five conditions included in the newborn screening panel.

As of December 2011, a total of 3,664 NBS facilities (birthing centers, lying-in clinics, rural health units, infirmaries, secondary/secondary/tertiary hospitals) are offering NBS. These facilities educate parents about NBS during prenatal visits, perform blood sample collection for NBS, organize transport of samples to NSC, recall patients with positive screening results and assist in the referral and management of patients (NSRC 2011).

DOH is preparing guidelines for the implementation of an expanded NBS program in the country by 2013. There are ongoing negotiations for approval of fund allocation for the expanded newborn screening which will increase coverage from five disorders to more than 20 disorders. The panel will include hemoglobinopathies, amino acid disorders, organic acid disorders, and endocrine disorders.

Newborn hearing screening

Legislation for newborn hearing screening was passed in 2009. Republic Act 9709 (RA 9709; Padilla et al. 2009a) institutionalizes measures for the prevention and early diagnosis of congenital hearing loss among newborns, the provision of referral follow-up, recall and early intervention services to infants with hearing loss, counselling and other support services for families of newborns with hearing loss, to afford them all the opportunities to be productive members of the community. The highlights of the law are:

- DOH is the lead agency.
- Any healthcare practitioner who delivers, or assists in the delivery, of a newborn in the Philippines shall, prior to delivery, inform the parents or legal guardian of the newborn of the availability, nature and benefits of hearing loss screening among newborns or infants 3 months old and below.
- All newborns must have access to hearing loss screening.
- Health facilities must integrate hearing screening in its services.
- Establishment of a network among pertinent government and private sector stakeholders for policy development, implementation, monitoring, and evaluation to promote universal newborn hearing screening program in the country.
- Establishment and maintenance of a newborn hearing screening database.

- Inclusion of the newborn hearing screening fee in the insurance benefits; and
- Establishment of a Newborn Hearing Screening Reference Center.

Genetic service facilities in the country and abroad

There are limited facilities in the country that provide genetic services to the public and/or private sectors, as listed in Table 6. For tests with limited requests in the country, samples from patients are sent overseas for further testing.

Future development of medical genetic services in the Philippines

The future of genetic services is dependent on many factors. Limited attention is provided by government since the focus is still on eradication of infectious diseases that predominate the top ten causes of infant mortality and infant morbidity. Learning from the developed countries where eradication of infections eventually paved the way to improvement of genetic services, the Philippines must prepare now by giving more attention (in terms of budget and program planning) to genetic disorders.

The situation is aggravated by the lack of trained personnel to handle the patients with genetic diseases. There are only eight trained clinical geneticists and no genetic counsellors.

Medical genetic services are limited to the major cities — Manila, Cebu City and Davao City. Despite the presence of a fellowship program on Clinical Genetics, it is not a popular choice and competes with other pediatric subspecialties, i.e., infectious diseases, cardiology, pulmonology, gastroenterology, etc. The offering of a Master of Science in Genetic Counselling is a strategy to bridge the gap of clinical geneticists. DOH must take an active role in identifying health personnel who can be trained for the clinical genetics and genetic counselling programs to be fielded at the regional and provincial levels.

Table 6 Facilities providing genetic services in the country

Laboratory/service	Number	
	Public	Private
Newborn screening	3	1
Molecular	1	2
Biochemical	1	0
Cytogenetics	2	2
DNA analysis (paternity testing)	3	1

The integration of NBS in the public health system is an opportunity for introduction of genetic services in the 17 regions of the country. Currently, there are regional DOH NBS coordinators assisted by a full-time NBS nurse coordinators, who are also involved in other genetics-related activities, such as the Birth Defects Surveillance (BDS) and the Telegenetics Referral system. The BDS is piloted in 81 sites throughout the country and provides a template for reaching out to patients in remote areas (Padilla et al. 2011a).

Recognizing that 50 % of the population is in the pediatric age group, DOH has joined hands with IHG-NIH in the establishment of the VYLH composed of adolescents and young adults promoting an increased awareness among the youth as regards healthy lifestyle and healthy pregnancies. The VYLH-Philippines is a network of leaders from different youth organizations based in universities and communities in the country, and was organized with the aim of mobilizing the youth towards health work. Currently, the network is doing advocacy and promotional work in their respective schools and communities, focusing on (1) the significance of folic acid supplementation in the prevention of birth defects; (2) the promotion of newborn screening; and (3) lobbying for public support for the urgent passage of the Rare Disease Act (Padilla et al. 2012).

The *barangay* health workers, midwives and nurses remain as front-liners in the field. To successfully bring down the knowledge to the grassroots, this new information about genetic disorders must be included in the training of these health workers.

The establishment of an office for genetic diseases at the DOH may fast track the program planning for these patients and their families.

Access to genetic services

There is a national health insurance program in the country, but there is much debate on the percentage of families covered as well as the percentage of health expenses covered by government. In general, 48.4 % of health expenditure are still out-of-pocket expenses. Among the genetic tests, NBS is the only one covered by the national health insurance. All other expenses related to treatment upon diagnosis are out-of-pocket expenses for the family.

The following are barriers to accessing genetic services in the Philippines: (1) financial, since most families cannot afford out-of-pocket expenses for the expensive genetic testing and treatment; (2) geographical, being an archipelago of 7,107 islands; (3) lack of awareness among different stakeholders, i.e., health professionals and parents; (4) compromised access to genetic services at the regional and

provincial level; and (5) lack of geneticists and genetic counsellors.

State of genetic services

Human resources and training

In terms of human resources, there has been an increasing trend in the density of health workers in the country. There are 12 physicians, 61 nurses and 18 health workers per 10,000 individuals. There are still areas in the Philippines without doctors and nurses because there is a preference to live in the urban areas primarily because of conveniences for families, i.e., education for children (WHO 2009; PRC 2011). There are only eight trained geneticists, six of whom are in Metro Manila while two are practising in the provinces (one in Davao City and one in Cebu City) making a medical geneticist to population density ratio of about 1:11,751,625.

Genetics is taught primarily in medical school as topics integrated in Biochemistry, Pediatrics, Internal Medicine and Obstetrics (CHED 2006). For pediatrics, the Philippine Pediatric Society has included genetics as a core topic in its curriculum for all medical schools. To date, all clinical geneticists are pediatricians.

Medical genetics is a recognized specialty since year 2000. There is no separate department of Medical Genetics. Only the Department of Pediatrics at the PGH has a separate section for Clinical Genetics.

The Department of Pediatrics, PGH offers a 2-year fellowship program in Clinical Genetics. It is designed to provide broad clinical exposure to areas of dysmorphology, biochemical genetics, cytogenetics, molecular genetics, and neonatal screening programs. Components of the training program are genetics and metabolic clinics, ward rounds and participation in regularly scheduled pre-clinic and post-clinic conferences. Experience in genetic counselling and training in laboratory procedures for the diagnosis of genetic disorders are likewise included (Padilla 2008).

Formal post-graduate training programs for molecular genetics are also available in the country. A Master of Science in Molecular Biology and Biotechnology and a Doctor of Philosophy in Molecular Biology and Biotechnology are currently offered at UP Diliman; and a Master of Science in Molecular Medicine is currently offered in SLMC. However, formal post-graduate training programs for biochemical genetics and cytogenetics are not yet available in the country. In-house trainings and seminars are carried out by the faculty and senior laboratory scientists of the IHG-NIH.

The Department of Pediatrics, PGH recently started a 2-year Master of Science in Genetic Counselling (MSGC)

program in 2011, catering to nurses, doctors and other allied health professionals. The MSGC program is a response to the growing need for counselling services of two major DOH programs — the NBS program and the BDS program. In this program, students will have an in-depth understanding of the various genetic conditions, their clinical management, inheritance pattern, and prognosis. As the students complete the 2-year program, they will gain competency in providing psychosocial support to the patient and members of their families. Their training will allow them to (1) apply the basics of human genetics and the principles of medical genetics and genetic counselling to given clients; (2) provide supportive genetic counselling to families, serve as patient advocates, and refer individuals and families to community and/or local government support services; (3) serve as educators and resource for other health care professionals and for the general public; and 4) plan, develop and evaluate genetic services programs (Laurino et al. 2011).

In general, the field of Clinical Genetics is not a popular profession at this time. Majority of the original staff of the IHG-NIH have been absorbed by genetic laboratories overseas. The brain drain has affected the very limited number of geneticists and genetic laboratory staff.

Workload

Only PGH offers comprehensive clinical genetics services in the country. The core staff of IHG-NIH is manned by four clinical geneticists, three PhD molecular geneticists, molecular biologists, medical technologist and nurses. On an average, there are about 100 consultations per geneticist every year. In terms of laboratory workload in 2011, there were 1,493 chromosomal analyses, 136 DNA tests and 432 biochemical tests performed at the IHG.

All NBS laboratories are considered public health laboratories even if one NSC is located in a private hospital. Implementing guidelines are issued by DOH. Licensing and accreditation are also handled by DOH. The 2011 workload for the NBS laboratories are: NSC-NIH had 290,605 patients; NSC-Central Luzon had 141,425 patients; NSC-Visayas had 145,592 patients; and NSC-Mindanao had 135,958 patients.

Aside from the IHG and the NBS laboratories, most of the other laboratories offering genetic services are private/commercial, serving only a very small portion of the population.

Quality assurance of medical genetic services

The Cytogenetics Laboratory IHG-NIH participates at the Cytogenetic European Quality Assessment (CEQA).

NSCs undergo initial accreditation and a re-accreditation every 3 years. The NSCs avail of the quality control samples

biannually and proficiency testing samples quarterly from the Center for Disease Control and Prevention (CDC), Atlanta, GA, USA, for CH (TSH), CAH (17-OHP), PKU (Phe), GAL (galactose and galactose-1-phosphate); and Quality Assurance Program every 2–3 months for G6PD Deficiency from the Veterans General Hospital in Taipei, Taiwan.

National policies and legal frameworks

There are no specific national guidelines for medical genetic services. Only newborn screening is supported by legislation (Newborn Screening Act of 2004 or RA 9288) which includes sanctions for failure of the health facility to achieve the target coverage set by DOH (NSRC 2011).

Research priorities in genetics/genomics

Genetics and genomics compete with other disciplines in securing funding for research grants from the government.

IHG-NIH is working on several collaborative projects. Some of the major projects are: (1) BDS and telegenetics referral system in collaboration with the DOH under the MoD-GNMIH; (2) the Subphenotyping and Genetics in Oral–Facial Cleft Families in the Philippines Project in collaboration with the University of Iowa and University of Pittsburgh; (3) pre-conception project with DOH and Lipa City Health Office under the MoD-GNMIH; and (4) Pan-Asian Single Nucleotide Polymorphism Initiative, a collaborative project between the IHG-NIH and the National Commission for Indigenous Populations (NCIP).

A recent development was the establishment of the PGC under the UP System in 2009. Research grants are made available for specific programs on health, biodiversity, agriculture, ethnicity and forensics, and ethical, legal and social aspects. In health, the NIH takes the lead with IHG and the Institute of Molecular Biology and Biotechnology (IMBB) receiving research grants. Initial research grants are allocated to infectious diseases (dengue diagnostics and biomarkers for severity; H1N1 molecular surveillance) and complex diseases (diabetes, cardiovascular diseases, cancers). Funds are being allocated by the Department of Science and Technology (Philippine Council for Health Research and Development; Philippine Council for Industry and Energy Research and Development) and intramural grants of the UP System, NIH Philippines and IHG-NIH (PGC 2011).

Patient organizations and public education in genetics

Geneticists partner with parent/patient organizations for genetic disorders. Geneticists provide lectures to parents and other health professionals for a better understanding of the genetic disorders and its impact to the patient, the family and society in general.

Geneticists and parent groups join hands in lobbying with government offices (i.e., DOH, Department of the Interior and Local Government, Department of Social Welfare and Development and PHIC) for better access and benefits for the concerned population of patients.

Down Syndrome Association of the Philippines, Inc. (DSAPI), a non-stock, non-profit organization, was established in 1992 by a group of parents and physicians to offer support to families who have a child with Down syndrome. The organization offers parent counselling, seminars, lectures, free medical and dental clinic services, sports activities, and art activities. It also organizes educational trips to various parks, museums and schools and other social activities. Through the efforts of the parents, the month of February was declared as the “National Down Syndrome Consciousness Month” by the former President of the Philippines Gloria Macapagal-Arroyo (Proclamation No. 157, February 2002). With this proclamation, several government agencies (DOH, Department of Education, Department of Labor and Employment, Department of Social Welfare and Development, other related agencies and appropriate non-government organizations) were enjoined to support and cooperate with the activities of the DSAPI. One of the highlights of the annual celebration is the Happy Walk, a walkathon participated in by families of children with Down syndrome, professionals, government and other non-government supporters. This activity was simultaneously conducted in Manila (Luzon), Cebu (Visayas) and Davao (Mindanao) to promote Down Syndrome Awareness nationwide (DSAPI 1992).

The Philippine Society of Orphan Disorders (PSOD), a non-stock, non-profit organization, was founded in June 2006 to serve as a central network for the advocacy and effective coordination of all viable efforts to sustain a better quality of life for the individuals with orphan or rare disorders in the Philippines. The goals of PSOD are: (1) to increase public awareness about rare disorders in the Philippines; (2) to develop and establish a nationwide registry of relevant and material statistical information, medications, drug trials and all other pertinent information about orphan disorders; (3) to assist patients and their families, family support groups, doctors and researchers in the management of these conditions; (4) to promote and support relevant and timely researches for a better understanding and treatment of orphan disorders; (5) to participate in policy formulation, advocacy and legislations of national and international

relevance about orphan disorders; (6) to develop and strengthen the relationship among institutions that are involved in the care of individuals with orphan disorders; (7) to develop and provide training for parents, families, health professionals, paramedical personnel and community health workers on the care of patients with orphan disorders; (8) to develop member's core competencies in the management of orphan disorders; and (9) to be financially self-reliant and to mobilize resources in the pursuit of organizational objectives. Members of the organization are health professionals, geneticists and pediatricians, parents, child advocates and concerned citizens. PSOD is currently lobbying for the passage of the Rare Disease Act of the Philippines, a bill that seeks to establish a system that will help ensure the early diagnosis and treatment of rare diseases in the Philippines (PSOD 2006).

Currently, the patients and families with the following conditions are being supported by the organization: urea cycle disorders (arginosuccinate lyase deficiency, carbamoyl phosphate synthetase [CPS] deficiency, ornithine transcarbamylase [OTC] deficiency, citrullinemia); organic acid disorders (methylmalonic aciduria [MMA]); amino acid disorders (phenylketonuria [PKU], maple syrup urine disease [MSUD], homocystinuria [HCY], tyrosinemia [TRY]); disorders of carbohydrate metabolism (glycogen storage disease, galactosemia [Gal]); lysosomal storage disease (mucopolysaccharidosis I [MPS I/Hurler/Hurler-Scheie/Scheie, mucopolysaccharidosis II [MPS II/Hunter syndrome], Sanfilippo syndrome, Morquio syndrome, Pompe disease, Gaucher disease, Batten disease, Fabry disease, Krabbe disease, Niemann–Pick disease, and Tay–Sachs disease); leukodystrophy (adrenoleukodystrophy [ALD]); and others (Chargé syndrome, Wilson disease).

Balikatang Thalassaemia is a non-stock, non-profit, non-political corporation founded in 1995 with a main objective of providing medical assistance, education, and counselling to patients with thalassaemia, their parents, and loved ones.

Conclusion

Being a developing middle-income country, the Philippines is faced with the challenge of providing healthcare for all Filipinos. Although one of the most active countries in Southeast Asia with regard to genetics, the country still has a shortage of geneticists and genetic counsellors. Difficulties exist for continued research and integration of healthcare services into the public health system. Geographical, cultural and financial barriers must be hurdled by the country in order to realize this goal. However, despite these shortcomings, the IHG-NIH views a promising future for medical genetics in the country, with the help of the government and support of the community.

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References

- Administrative Order (AO) 2002-0096 – Guidelines for multiple micronutrient supplementation for pregnant women in UNICEF CPC V Areas. <http://home.doh.gov.ph/ao/ao96-02.pdf>. Accessed 17 January 2012
- Administrative Order (AO) 2003-0119 — Updated guidelines on micronutrient supplementation (vitamin a, iron, and iodine). <http://home.doh.gov.ph/ao/ao119-03.pdf>. Accessed 17 January 2012
- Administrative Order (AO) 2010-0010 Revised policy on micronutrient supplementation to support achievement of 2015 MDG targets to reduce under five and maternal deaths and address micronutrient needs of other population groups. http://202.90.151.88/nnc/opt_cgs/dohao10.pdf. Accessed 17 January 2012
- Briones TK, Tantengco AT, Pascasio FM, Garcia FP, Reyes MT (eds) (1984) Applied human genetics. JMC Press, Inc., Quezon City, Philippines, pp 145–154
- Central Intelligence Agency (CIA) (2011) The world factbook. <https://www.cia.gov/library/publications/the-world-factbook/geos/rp.html>. Accessed 17 January 2012
- Christianson A, Howson CP, Modell B (2006) March of Dimes (MoD) Global report on births defects, 2006: the hidden toll of dying and disabled children. http://www.marchofdimes.com/professionals/871_18587.asp. Accessed 17 January 2012
- Commission on Higher Education (CHED) (2006) CHED Memorandum order No. 10 series of 2006: policies, standards and guidelines for medical education. www.ched.gov.ph/chedwww/index.php/eng/content/download/686/3849/file/CMO_10_S_2006.pdf. Accessed 17 January 2012
- Cutiongco-de la Paz EM (2006) Prenatal diagnosis and its role in reproductive risk screening, prevention and treatment of genetic diseases: is the Philippines ready for it? *Acta Med Philipp* 40(2):54–57. Available at www.actamedicaphilippina.com.ph
- Department of Health, Republic of the Philippines Statistics (DOH) (2006) Statistics: Top 10 causes of infant mortality. http://www.doh.gov.ph/kp/statistics/infant_deaths. Accessed 17 January 2012
- Department of Health, Republic of the Philippines (DOH) (1999) Sa reporma... health ang una! Health Sector Reform Agenda. DOH, Manila, pp 1999–2004
- Dolan RE (ed) (1991) Philippines: a country study. Washington: GPO for the Library of Congress. <http://countrystudies.us/philippines/>. Accessed 17 January 2012
- Down Syndrome Association of the Philippine, Inc. (DSAPI) (1992) <http://www.dsapi.org>. Accessed 17 January 2012
- International Monetary Fund (IMF) (2011) World economic and financial surveys, world economic outlook database. <http://www.imf.org/external/pubs/ft/weo/2011/02/weodata/index.aspx>. Accessed 17 January 2012

- Laurino MY, Padilla CD, Alcausin MMB, Silao CLT, Cutiongco-de la Paz EM (2011) A master of science in genetic counseling program in the Philippines. *Acta Med Philipp* 45(4):7–9. Available at www.actamedicaphilippina.com.ph
- March of Dimes (MoD) (2012) Global network for maternal and infant health. http://www.marchofdimes.com/mission/globalprograms_gnmih.html. Accessed 17 January 2012
- Murray JC, Daack-Hirsch S, Buetow KH et al (1997) Clinical and epidemiologic studies of cleft lip and palate in the Philippines. *Cleft Palate Craniofac J* 34(1):7–10
- National Council for Disability Affairs (NCDA) (2011) <http://ncda.gov.ph>. Accessed 17 January 2012
- National Statistics Office (NSO) (1998) Republic of the Philippines, Department of Health (DOH), Republic of the Philippines and Macro International Inc. (MI). National Demographic and Health Survey 1998. Manila: National Statistics Office and Macro International Inc. http://pdf.usaid.gov/pdf_docs/PNACF095.pdf. Accessed 17 January 2012
- National Statistics Office (NSO) (2003) Republic of the Philippines, and ORC Macro. National Demographic and Health Survey 2003. Calverton, Maryland: NSO and ORC Macro. <http://rochr.qrc.com/bitstream/123456789/1590/1/FR156.pdf>. Accessed 17 January 2012
- National Statistics Office (NSO) (2008) Republic of the Philippines, and ICF Macro. National Demographic and Health Survey 2008. Calverton, Maryland: National Statistics Office and ICF Macro. <http://www.measuredhs.com/pubs/pdf/FR224/FR224.pdf>. Accessed 17 January 2012
- National Statistics Office (NSO) (2009) Republic of the Philippines. The Philippines in Figures 2009. http://www.census.gov.ph/data/publications/pif_2009.pdf. Accessed 17 January 2012
- Newborn Screening Reference Center (NSRC) (2011) <http://www.newbornscreening.ph/>. Accessed 17 January 2012
- Padilla CD (2008) Overview of genetic health services at UP Manila. *Acta Med Philipp* 42(2):7–10. Available at www.actamedicaphilippina.com.ph
- Padilla CD, Cutiongco EM, Sia JM (2003) Birth defects ascertainment in the Philippines. *Southeast Asian J Trop Med Public Health* 34 (Suppl 3):239–343
- Padilla CD, Cutiongco-de la Paz EM, Lucero F et al (2008) for the Philippine Oral Cleft Registry Study Group. Profile of oral cleft cases reported in the Philippine Oral Cleft Registry from May 2003 to December 2006. *Acta Med Philipp* 42(2):27–33. Available at www.actamedicaphilippina.com.ph
- Padilla CD, Basilio JA, Oliveros YE (2009a) Newborn screening: research to policy. *Acta Med Philipp* 43(2):6–14. Available at www.actamedicaphilippina.com.ph
- Padilla CD, Dans L, Tamondong M, Bernal R, Lacey JJ and Estrada S (2009b) Cost–benefit analysis of the Newborn Screening Program of the Philippines. *Acta Med Philipp* 43(2):46–52. Available at www.actamedicaphilippina.com.ph
- Padilla CD, de la Paz EMC, Cavan BC et al (2011a) Establishment of the Philippine Birth Defects Surveillance. *Acta Med Philipp* 45(4):12–19. Available at www.actamedicaphilippina.com.ph
- Padilla CD, Berbos AGD, Abadingo M et al (Padilla 2011b) Occurrence of birth defects at the Philippine General Hospital: 2001–2010. *Acta Med Philipp* 45(4):20–29. Available at www.actamedicaphilippina.com.ph
- Padilla CD, de la Paz EMC, Silao CL (2012) Medical Genetic Services in the Philippines. In: Kumar D (ed) *Genomics and health in the developing world*. Oxford University Press, New York, pp 816–826
- Philippine Genome Center (PGC) (2011) <http://pgc.up.edu.ph/>. Accessed 17 January 2012
- Philippine National Health Accounts (PNHA) (2005) National Statistical Coordination Board. <http://www.nscb.gov.ph/stats/pnha/2005/default.asp>. Accessed 17 January 2012
- Philippine Society for Orphan Disorders (PSOD) (2006) <http://www.psod.org.ph/ver2/>. Accessed 17 January 2012
- Presidential Proclamation No. 1188 (2006) <http://home.doh.gov.ph/mc/mc2008-0006.pdf>. Accessed 17 January 2012
- Professional Regulation Commission (PRC) (2011) Republic of the Philippines. Official website. <http://www.prc.gov.ph/prb/default.aspx>. Accessed 17 January 2012
- Republic Act (RA) 8172 (1995) An Act for Salt Iodization Nationwide (ASIN). Population and family planning, laws, policies and regulations. http://www.unescap.org/esid/psis/population/database/poplaws/law_phi/phi_009.htm. Accessed 17 January 2012
- Republic Act (RA) 8976 (2000) The Philippine Food Fortification Act of 2000 Arellano University Foundation. The LawPhil Project: RA8976. http://www.lawphil.net/statutes/repacts/ra2000/ra_8976_2000.html. Accessed 17 January 2012
- Republic Act (RA) 9442 (2006) An act amending the Magna Carta for Persons with Disability (RA 7277) National Council on Disability Affairs. <http://www.ncda.gov.ph/disability-laws/implementing-rules-and-regulations-irr/implementing-rules-and-regulations-of-republic-act-no-9442/>. Accessed 17 January 2012
- Republic Act (RA) 9709 – The Universal Newborn Hearing Screening and Intervention Act of 2009 Arellano University Foundation. The LawPhil Project: RA9709. http://www.lawphil.net/statutes/repacts/ra2009/ra_9709_2009.html. Accessed 17 January 2012
- Rubella Campaign (2011) Department of Health, Republic of the Philippines. <http://www.doh.gov.ph/content/iligtas-sa-tigdasang-%E2%80%98pinas>. Accessed 17 January 2012
- United Nations Development Programme (UNDP) (2009) Human development report. Overcoming barriers: human mobility and development. <http://hdr.undp.org/en/reports/global/hdr2009/>. Accessed 17 January 2012
- United Nations Educational, Scientific and Cultural Organization (UNESCO) (2009) UNESCO National Education Support Strategy (UNESS): Philippines. www.unesconatcom.ph/docs/education/uness_report.pdf. Accessed 17 January 2012
- World Health Organization (WHO) (2009) World Health Statistics. <http://www.who.int/whosis/whostat/2009/en/index.html>. Accessed 17 January 2012